

# Leukaemia Section

## Short Communication

### t(12;14)(p13;q32)

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## Clinics and pathology

### Disease

Acute lymphoblastic leukemia.

### Phenotype / cell stem origin

Pre B ALL.

### Epidemiology

Only one case to date; no age nor gender data.

### Prognosis

No data.

## Cytogenetics

### Cytogenetics molecular

Cryptic translocation; recognized by multicolor FISH plus locus-specific FISH.

### Additional anomalies

Hyperploidy (57 chromosomes).

## Genes involved and Proteins

### ETV6

**Location:** 12p13

### Protein

53 and 57 kDa; contains, from N-term to C-term, an helix-loop-helix (HLH) domain (or 'sterile alpha domain' SAM) responsible of dimerization, and a sequence specific DNA-binding domain (ETS domain); binds to 5' CCGGAAGT 3'; ETV6 is a member of the ETS family of transcription factors; transcriptional repressor.

### IGH

**Location:** 14q32

## References

Lu XY, Harris CP, Cooley L, Margolin J, Steuber PC, Sheldon M, Rao PH, Lau CC. The utility of spectral karyotyping in the cytogenetic analysis of newly diagnosed pediatric acute lymphoblastic leukemia. *Leukemia* 2002;16:2222-2227.

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